



## RPL5 gene

ribosomal protein L5

### Normal Function

The *RPL5* gene provides instructions for making one of approximately 80 different ribosomal proteins, which are components of cellular structures called ribosomes. Ribosomes process the cell's genetic instructions to create proteins.

Each ribosome is made up of two parts (subunits) called the large and small subunits. The protein produced from the *RPL5* gene is among those found in the large subunit.

The specific functions of the RPL5 protein and the other ribosomal proteins within these subunits are unclear. Some ribosomal proteins are involved in the assembly or stability of ribosomes. Others help carry out the ribosome's main function of building new proteins. Studies suggest that some ribosomal proteins may have other functions, such as participating in chemical signaling pathways within the cell, regulating cell division, and controlling the self-destruction of cells (apoptosis).

### Health Conditions Related to Genetic Changes

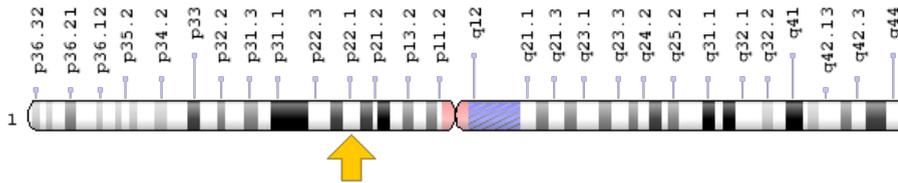
#### Diamond-Blackfan anemia

More than 30 *RPL5* gene mutations have been identified in individuals with Diamond-Blackfan anemia. These mutations are believed to affect the stability or function of the RPL5 protein. Studies indicate that a shortage of functioning ribosomal proteins may increase the self-destruction of blood-forming cells in the bone marrow, resulting in a low number of red blood cells (anemia). Abnormal regulation of cell division or inappropriate triggering of apoptosis may contribute to the other health problems and unusual physical features that affect some people with Diamond-Blackfan anemia.

## Chromosomal Location

Cytogenetic Location: 1p22.1, which is the short (p) arm of chromosome 1 at position 22.1

Molecular Location: base pairs 92,832,037 to 92,841,924 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 60S ribosomal protein L5
- DBA6
- L5
- MGC117339
- MSTP030
- PPP1R135
- RL5\_HUMAN

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The RNA message is decoded on ribosomes  
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1071>

### GeneReviews

- Diamond-Blackfan Anemia  
<https://www.ncbi.nlm.nih.gov/books/NBK7047>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RPL5%5BTIAB%5D%29+OR+%28ribosomal+protein+L5%5BTIAB%5D%29%29+OR+%2860S+ribosomal+protein+L5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- RIBOSOMAL PROTEIN L5  
<http://omim.org/entry/603634>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_RPL5.html](http://atlasgeneticsoncology.org/Genes/GC_RPL5.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=RPL5%5Bgene%5D>
- Diamond-Blackfan Anemia Mutation Database  
[http://www.dbagenes.unito.it/home.php?select\\_db=RPL5](http://www.dbagenes.unito.it/home.php?select_db=RPL5)
- HGNC Gene Family: L ribosomal proteins  
<http://www.genenames.org/cgi-bin/genefamilies/set/729>
- HGNC Gene Family: Protein phosphatase 1 regulatory subunits  
<http://www.genenames.org/cgi-bin/genefamilies/set/694>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=10360](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10360)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6125>
- UniProt  
<http://www.uniprot.org/uniprot/P46777>

## Sources for This Summary

- Boria I, Garelli E, Gazda HT, Aspesi A, Quarello P, Pavesi E, Ferrante D, Meerpohl JJ, Kartal M, Da Costa L, Proust A, Leblanc T, Simansour M, Dahl N, Fröjmark AS, Pospisilova D, Cmejla R, Beggs AH, Sheen MR, Landowski M, Buros CM, Clinton CM, Dobson LJ, Vlachos A, Atsidaftos E, Lipton JM, Ellis SR, Ramenghi U, Dianzani I. The ribosomal basis of Diamond-Blackfan Anemia: mutation and database update. *Hum Mutat.* 2010 Dec;31(12):1269-79. doi: 10.1002/humu.21383.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20960466>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4485435/>
- Cmejla R, Cmejlova J, Handrkova H, Petrak J, Petrtlyova K, Mihal V, Stary J, Cerna Z, Jabali Y, Pospisilova D. Identification of mutations in the ribosomal protein L5 (RPL5) and ribosomal protein L11 (RPL11) genes in Czech patients with Diamond-Blackfan anemia. *Hum Mutat.* 2009 Mar;30(3):321-7. doi: 10.1002/humu.20874.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19191325>
- Ellis SR, Gleizes PE. Diamond Blackfan anemia: ribosomal proteins going rogue. *Semin Hematol.* 2011 Apr;48(2):89-96. doi: 10.1053/j.seminhematol.2011.02.005. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21435505>
- Farrar JE, Dahl N. Untangling the phenotypic heterogeneity of Diamond Blackfan anemia. *Semin Hematol.* 2011 Apr;48(2):124-35. doi: 10.1053/j.seminhematol.2011.02.003. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21435509>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3078697/>
- Gazda HT, Sheen MR, Vlachos A, Choessel V, O'Donohue MF, Schneider H, Darras N, Hasman C, Sieff CA, Newburger PE, Ball SE, Niewiadomska E, Matysiak M, Zaucha JM, Glader B, Niemeyer C, Meerpohl JJ, Atsidaftos E, Lipton JM, Gleizes PE, Beggs AH. Ribosomal protein L5 and L11 mutations are associated with cleft palate and abnormal thumbs in Diamond-Blackfan anemia patients. *Am J Hum Genet.* 2008 Dec;83(6):769-80. doi: 10.1016/j.ajhg.2008.11.004.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19061985>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668101/>
- Ito E, Konno Y, Toki T, Terui K. Molecular pathogenesis in Diamond-Blackfan anemia. *Int J Hematol.* 2010 Oct;92(3):413-8. doi: 10.1007/s12185-010-0693-7. Epub 2010 Sep 30. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20882441>
- Lipton JM, Ellis SR. Diamond-Blackfan anemia: diagnosis, treatment, and molecular pathogenesis. *Hematol Oncol Clin North Am.* 2009 Apr;23(2):261-82. doi: 10.1016/j.hoc.2009.01.004. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19327583>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2886591/>
- Narla A, Hurst SN, Ebert BL. Ribosome defects in disorders of erythropoiesis. *Int J Hematol.* 2011 Feb;93(2):144-9. doi: 10.1007/s12185-011-0776-0. Epub 2011 Feb 1. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21279816>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3689295/>
- Quarello P, Garelli E, Carando A, Brusco A, Calabrese R, Dufour C, Longoni D, Misuraca A, Vinti L, Aspesi A, Biondini L, Loreni F, Dianzani I, Ramenghi U. Diamond-Blackfan anemia: genotype-phenotype correlations in Italian patients with RPL5 and RPL11 mutations. *Haematologica.* 2010 Feb;95(2):206-13. doi: 10.3324/haematol.2009.011783. Epub 2009 Sep 22.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19773262>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2817022/>
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<http://omim.org/entry/603634>

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